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A different type of hyperlipidemia

Giovanna Lovatón Villena(1), A Guarido(1), E Gregorutti(1), C García(2)
(1) Family Practice, Consorci Sanitari de Terrassa, Spain
(2) Rheumatology Department, Consorci Sanitari de Terrassa, Spain

Corresponding author: Dr Giovanna Lovatón, Consorci Sanitari de Terrassa, Cap Est, Terrassa, Spain. E-mail: giovannalv@hotmail.com

Backgroud & Aim: The glycogen-storage diseases (GSD) are caused by enzymatic defects of glycogen. Their global incidence is 1: 20000 newborns. The deficiency of the G6Pase causes GSD type Ia, the defect in glucosa-6-phosphate translocate (transporter) causes GSD type Ib and deficiency of the debranching enzyme GSD type III, which is measured in muscle and liver biopsy.
Method: 54 year old woman is seen for the first time in our primary care center for arthralgia. As medical relevant history, glycogenesis which was followed in another hospital until the age of 17, was not provided any clinical history. She remembers that a liver biopsy was held. Physical examination revealed swelling and pain in the right second metacarpal phalanx and in the right metatarsophalangeal area.
Results: Laboratory findings were leukopenia, hyperuricemia, hypercolestherolemia, hypertriglyceridemia, rheumatoid factor in 69,2 and hyperferritinemia. A consultation with rheumatology and after the assessment, laboratory tests in three months showed normalization of leucopenia, normal glucose-6-phosphatase (G6Pase) also normal x-ray of hands and feet. Prescribed celecoxib for the pain. They also made further consultation with Gastroenterology, who requested abdominal ultrasound because persistent hyperferritinemia with normal liver enzymes, still pending. We also did a consultation with internal medicine and nutrition for the multidisciplinary management.
Conclusions: GSD type Ia is the most frequent GSD and initial symptoms are due to hypoglicemia, liver may be enlarged at birth. With ageing the patient may present with poor growth, short stature as in our patient. In rare individuals with milder clinical manifestations, G6Pase enzyme activity can be higher. Laboratory abnormalities in addition to hypoglicemia are, lactic acidosis, hyperlipidemia (particularly hypertriglyceridemia) and hyperuricemia. Kidneys may be enlarged. Without effective treatment, long-term complications occur, namely hepatic adenomas, renal dysfunction and urolithiasis, osteoporosis and gout. Hyperlipidemia may cause xanthomas, pancreatitis and cholelitiasis. It is recommended abdominal ultrasonography with alpha fetoprotein and carcinoembryonic antigen levels every 3 months once patients develop hepatic lesions. Despite increased levels of triglyceride, VLDL and LDL in GSD-Ia patients, endothelial vascular dysfunction and atheroesclerosis are rare.